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Journal of Pediatric Surgery CASE REPORTS

journal homepage: www.jpascasereports.com

Concurrent esophageal atresia with tracheoesophageal fistula and Hirschsprung disease

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ARTICLE INFO

Article history:

Received 6 July 2015

Received in revised form

21 September 2015

Accepted 24 September 2015

Key words:

Hirschsprung disease

Esophageal atresia and tracheoesophageal fistula

Congenital malformations

ABSTRACT

We describe two cases of concomitant Hirschsprung disease and esophageal atresia and tracheoesophageal fistula in the newborn, both of which were successfully diagnosed and managed in the neonatal period. This is the first report in the English literature to identify the coexistence of these distinct congenital malformations.

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Esophageal atresia \pm tracheoesophageal fistula (EA \pm TEF) occurs in 1/2500–4500 live births and is one of the VACTERL (vertebral, anorectal, cardiac, trachea-esophageal, renal and limb) associated malformations [1]. Hirschsprung disease (HD) occurs in 1/2000–5000 live births and is a physiologic defect caused by distal aganglionosis of bowel that leads to spasm and symptoms of distal intestinal obstruction [2]. Although some suggest that HD is associated with anorectal malformations [2] and EA \pm TEF is associated with VACTERL, the co-occurrence of HD and TEF malformations is currently unreported in the English literature.

1. Case A

A 39 week male, born to a 22 year old gravida 1, para 1, developed copious secretions and labored respirations after birth at an outside facility. A chest radiograph confirmed a Replogle tube coiled in the esophageal pouch. The patient was then transferred to our institution on day of life (DOL) 1 for management of presumed EA + TEF. His subsequent VACTERL workup was negative including

echocardiogram, renal ultrasound, spinal ultrasound and skeletal radiographs.

On DOL 2, intraoperative bronchoscopy identified a Type C EA + TEF, which was followed by an uncomplicated, routine operative repair including chest tube placement. On post-operative day (POD) 2, the patient developed abdominal distension and bilious emesis. Abdominal radiograph demonstrated a distal bowel obstruction, and a subsequent contrast enema revealed a narrow rectum and proximal caliber change at the sigmoid colon concerning for HD. A suction rectal biopsy was performed and rectal irrigations were initiated. Despite colonic irrigations his abdominal distension persisted and he quickly developed a pneumothorax. Due to this rapid progression of symptoms and patient instability, he was taken to the operating room for placement of an additional chest tube and creation of a diverting, end ileostomy and mucus fistula. Ileostomy was chosen due to the urgency of the procedure, we did not want to wait for frozen section results and it allows for a protective stoma at the time of the definitive pull-through. After improving the drainage of the pneumothorax in conjunction with decompression of the abdominal cavity the patient's clinic status vastly improved. Full thickness biopsies were then performed and later confirmed the level of aganglionic disease at the distal descending colon, which was marked and directed the level of resection at the time of definitive repair. To further work-up his worsening

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pneumothorax, we evaluated his TEF repair with micro-laryngoscopy and bronchoscopy, which were negative, followed by esophagram on POD 7 that was without anastomotic leak.

At 7 weeks of age, the patient underwent definitive treatment of his HD by transanal (Swenson type) pull-through with rectosigmoid resection and coloanal anastomosis in addition to placement of a gastrostomy tube. His ileostomy was closed and he no longer requires gastrostomy tube feeds. The patient's post-operative course was uneventful. A pediatric geneticist concluded that no chromosomal anomaly was present given the lack of dysmorphism and lack of additional systemic involvement.

2. Case B

The second report of concurrent EA + TEF and HD was initially managed at another institution and referred to us for management of a post-operative rectal stricture. This 39 week gestation male was born to a woman with a history significant only for Herpes simplex virus infection at 10 weeks of prenatal care. Similarly, he presented with increased oropharyngeal secretions, respiratory distress, abdominal distension and inability to pass an orogastric tube. Chest radiograph confirmed a probable blind esophageal pouch with a Type C TEF due to distal intestinal gas. Upon transfer to a regional specialty center at 8 h of life, he underwent a VACTERL work-up that revealed normal renal and spinal anatomy. Echocardiography demonstrated non-surgical lesions including a small patent ductus arteriosus and an atrial level shunt that was either a patent foramen ovale or secundum atrial septal defect.

On DOL 2 he developed worsening abdominal distension followed by respiratory distress that required urgent surgery prior to TEF repair. A pre-operative contrast enema was consistent with distal colonic obstruction. During intubation, a rigid bronchoscopy was performed and a Fogarty catheter was inserted to obstruct the TEF. Following successful intubation, a laparotomy was performed demonstrating proximal colonic dilation and distal collapse consistent with HD. A colon biopsy was performed to confirm the diagnosis, leveling colostomy created and a gastrostomy tube was placed.

On DOL 4 he returned to the operating room for a primary repair of his EA and ligation of distal TEF. Enteral feeds were initiated after gastrografin swallow confirmed an intact esophageal anastomosis without leak.

Beside suction rectal biopsies on DOL 15 again confirmed the diagnosis of HD showing prominent submucosal nerves and aganglionosis. At 5 months of age, the patient underwent elective repair with a transanal endorectal (Soave) pull-through and colostomy takedown.

3. Discussion

In both cases of concomitant HD with EA + TEF, the term newborns were without significant comorbidities and were diagnosed with the most common subtype of EA + TEF, Type C. Initial presentation of both infants was similar and classic for the diagnosis of EA ± TEF [1]. The synchronous congenital malformations were identified soon after birth with HD symptomatically presenting DOL 4, 2 days post-repair of EA, in the first case and on DOL 2 pre-repair of EA in the second. Abdominal radiograph and contrast enema raised suspicion of HD in each case, and both were managed by an early fecal diversion procedure followed by transanal pull-through.

The co-occurrence of EA ± TEF and HD is currently unreported in the English literature. When considered individually, each anomaly has a higher incidence in Down syndrome (DS) patients with a prevalence of 0.3–0.8% (EA) and 0.6–3% (HD) [3,4]. We suspected that the simultaneous occurrence of both malformations would be very low in the general population but possibly have a higher prevalence in the DS population. However, the association is not reported in either the trisomy 21 or general population. To identify similar cases nationally, we queried the National Birth Defects Prevention Study, created by the Center for Disease Control and Prevention (USA), and found 5 unconfirmed cases of concurrent HD and EA ± TEF out of more than 7500 EA ± TEF and 4800 HD cases in the database [5–8]. Additionally we searched our institution's HD database of 334 patients and found no other cases of HD with EA. The incidence is too low to predict prevalence of this rare co-occurrence.

Although EA ± TEF and HD each have a higher incidence in DS, their coexistence is similar in both the DS and general populations [9,10]. Furthermore, this suggests that our cases, in the absence of dimorphisms and additional malformations, are likely spontaneous occurrences.

4. Conclusions

We conclude that this rare association of congenital EA + TEF and HD in otherwise healthy newborns is the first reported in the English literature. In both cases, EA + TEF was the initial anomaly diagnosed but HD was recognized shortly after, by DOL 4. After searching our institution's database and the National Birth Defects Prevention Study, we found only 5 possible, additional cases that were unconfirmed. The simultaneous occurrence of EA ± TEF and HD is extremely rare and previously unreported.

Conflict of interest

No conflict of interest or financial disclosures.

References

- [1] Pinheiro PF, Simoes e Silva AC, Pereira RM. Current knowledge on esophageal atresia. *World J Gastroenterol* 2012;18(28):3662–72. <http://dx.doi.org/10.3748/wjg.v18.i28.3662>.
- [2] Hofmann AD, Puri P. Association of Hirschsprung's disease and anorectal malformation: a systematic review. *Pediatr Surg Int* 2013;29(9):913–7. <http://dx.doi.org/10.1007/s00383-013-3352-2>.
- [3] Freeman SB, Torfs CP, Romitti PA, Royle MH, Druschel C, Hobbs CA, et al. Congenital gastrointestinal defects in Down syndrome: a report from the Atlanta and National Down Syndrome Projects. *Clin Genet* 2009;75(2):180–4. <http://dx.doi.org/10.1111/j.1399-0004.2008.01110.x>.
- [4] Kallen B, Mastroiacovo P, Robert E. Major congenital malformations in Down syndrome. *Am J Med Genet* 1996;65(2):160–6. [http://dx.doi.org/10.1002/\(SICI\)1096-8628\(19961016\)65:2<160::AID-AJMG16>3.0.CO;2-O](http://dx.doi.org/10.1002/(SICI)1096-8628(19961016)65:2<160::AID-AJMG16>3.0.CO;2-O).
- [5] National Birth Defects Prevention Network (NBDPN): congenital malformations surveillance report, 97; 2013. p. S1–172.
- [6] National Birth Defects Prevention Network (NBDPN): congenital malformations surveillance report, 79; 2007. p. 874–942.
- [7] Parker SE, Mai CT, Canfield MA, Rickard R, Wang Y, Meyer RE, et al. National Birth Defects Prevention Network. Updated National Birth Prevalence estimates for selected birth defects in the United States, 2004–2006. *Birth Defects Res A Clin Mol Teratol* 2010;88(12):1008–16. <http://dx.doi.org/10.1002/bdra.20735>.
- [8] Yoon PW, Rasmussen SA, Lynberg MC, Moore CA, Anderka M, Carmichael SL, et al. The National Birth Defects Prevention Study. *Public Health Rep* 2001; 116(Suppl. 1):32–40.
- [9] Martinez- Frias ML UM, Cereijo A. Anorectal and esophageal anomalies in Down syndrome. *Am J Med Genet* 1992;44:848–9.
- [10] Torfs CP, Bateson TF, Curry CJ. Anorectal and esophageal anomalies with Down syndrome. *Am J Med Genet* 1992;44(6):847. <http://dx.doi.org/10.1002/ajmg.1320440632>. author reply 848–850.